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Book Review

DEVELOPMENTAL DISABILITIES IN INFANCY AND CHILDHOOD, 2nd Ed., Vols. I, II.
Arnold J. Capute and Pasquale J. Accardo, Editors.
Paul H. Brookes Publishing, Baltimore, MD, 1996, 1159pp, $195.

It may be an exaggeration to describe developmental pediatrics and pediatric genetics as two different worlds. While functional assessment and management are primary concerns of developmentalists, these attributes are inseparable from the causal diagnoses emphasized by geneticists and neurologists. However, the expanded second edition by Capute and Accardo gives tacit support to the separation of developmental and genetics clinics at most medical centers. Their Foreword anticipates a union of developmental pediatrics and child neurology, but mentions little about genetics. Despite this neglect, they provide a valuable resource for medical geneticists.

As might be anticipated from the Foreword, the coverage of traditional genetics is quite selective. Volume I has pertinent chapters on the genetics of embryonic development (Roy Breg), metabolic disease (Richard Kelley), and the genetic examination (Thomas Montgomery and Kenneth Jones); volume II has chapters on Down (Paul Rogers, Nancy Roizen, George Capone), Prader-Willi (Vanja Holm), fragile X (Andrew Adesman), Williams (Barbara Pober), Rett (Hugo Moser and Sakkubai Naidu), and fetal alcohol (Mary Leppert, Karen Hofman) syndromes. A broader genetic perspective is served by chapters on visual impairment (Alexander Hoon), hearing loss (Nancy Roizen), and Tourette syndrome (Harvey Singer) in volume I.

What is included in genetics is very useful. The chapter on Down syndrome is an outstanding, concise summary of phenotypic/molecular genetic correlation, natural history, management, and psychosocial support. The chapters on Williams and Prader-Willi syndromes are also helpful in defining natural history and management, but these aspects are neglected in the chapter on fetal alcohol syndrome. The latter chapter and that on fragile X syndrome display a weakness in these volumes: figures are sparse and sometimes amateurishly composed (e.g., the cytogenetic and DNA data on fragile X syndrome), or poorly selected (e.g., the examples of fetal alcohol syndrome showing uncharacteristic manifestations of down-slanting palpebral fissures, bulbous nose, and a small mouth).

What is excluded from genetics coverage constitutes a more substantial weakness of the books. Roy Breg nicely summarizes the major genetic themes in development (DNA-binding motifs, cell signaling, apoptosis, sex determination, imprinting, triplet repeat expansion), but this chapter alone cannot do justice to the genetic triumphs of inheritance pattern, linkage, positional cloning, informative mutation, and genome map. Critical knowledge of family history, pedigree analysis, and cytogenetic or DNA diagnosis is not reviewed. More discussion of the approach to children with morphologic or metabolic disease is also needed, including additional examples of disease categories in the chapter on the clinical examination and better diagnostic algorithms in the chapter on metabolic disease. Chapters that began with a classical developmental phenotype (e.g., hyperactivity or cerebral palsy) and presented the appropriate genetic differential (e.g., San Filippo syndrome or the X-linked spastic diplegias) would be ideal, but such chapters might be difficult to entice from contributors.

Given that Capute and Accardo did not intend to provide a comprehensive review of genetics, how valuable are these volumes for the geneticist who wishes to become more familiar with developmental material? I believe they are tremendously useful, and strongly recommend them for this purpose. The world of developmental pediatrics is served in volume I by chapters on the epidemiology of developmental disabilities (Paul Lipkin), the developmental history (Frederick Palmer), and more than eight chapters on the neurodevelopmental assessment of children. Volume II provides chapters on cerebral palsy (four chapters), spina bifida and hydrocephalus (Peter Chauvel and Stephen Kinsman), childhood-acquired hydrocephalus (Stephen Kinsman), autism (three chapters), and disorders of learning or attention (five chapters).

Those geneticists not interested in classical developmental disorders such as autism will find other valuable subjects that are rarely included in medical genetics texts. Volume I includes chapters on neurologic development (three chapters), environmental causes (prematurity, congenital infections, AIDS, substance abuse, lead poisoning, traumatic brain injury), and the eight chapters on neurodevelopmental assessment. Volume II includes the medical history of developmental disabilities (Pasquale Accardo), fetal risk factors leading to developmental disabilities (Claudine Amiel-Tison), preterm neurosensory development (Marilee Allen), and the neuropathology of learning disorders (Walter Kaufmann).

Of even greater importance to clinical geneticists are the sections in volume I concerned with clinical care and social advocacy for children with developmental disabilities. Any health professional seeing children with mental disability should know about social supports, inclusive schooling, residential facilities, and legal issues (e.g., guardianship, special needs trusts). The chapters on interdisciplinary team process (Susan Farrell and Ada Pimentel), coping with developmental disabilities (Beverly Myers), medical issues in residential placement (Michael Kurtz and Wulfred Berman), legislative mandates (Robert Biehl), and transitions in

adolescents with disabilities (Chris Johnson) exemplify a few of the 12 chapters that concentrate on psychosocial issues.

In summary, the volumes by Capute and Accardo are worthwhile, even essential, for medical geneticists who are interested in management and psychosocial support after the initial diagnostic evaluation. There may be omissions from the developmental perspective that this geneticist reviewer did not recognize, but the experience and reputation of the authors in this area is endorsed in the bibliographies and by Forwords from Drs. Duane Alexander and Robert Cooke. Although the song "Two Different Worlds" ends with the hope that the two worlds will become one, Capute and Accardo provide an admirable but not quite successful attempt to realize this hope by uniting the worlds of developmental pediatrics and medical genetics. Their anticipation of "newly evolved developmental neuropediatricians" as an audience for the next edition may be realized, but the power of genetics should not be forgotten in the reach toward a medicine for the mind.

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